

THE NEURITIC TYPE OF PROGRESSIVE MUSCULAR ATROPHY. A CASE WITH MARKED HEREDITY.*

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It was just twenty years ago that Charcot and Marie gave a description of the neuritic type of progressive muscular atrophy, a description which remains classical to this day; to which very little has since been added and from which practically nothing has been taken away. In 1899 Sainton published a Paris thesis into which he gathered the majority of cases up to that time reported, and published an additional one of his own with full autopsy and histological examination. He carefully scrutinized all reported cases and exercised considerable latitude in excluding a number which might very properly have been accepted, among them one or two which had been submitted to post-mortem examination. Based upon his collected cases he describes the Charcot-Marie type as commonly hereditary and often familial, although occasionally occurring in isolated cases. It affects men much more frequently than women. Commonly it develops first in the lower extremities, sometimes in the upper, and occasionally in all four at once. In the lower extremities the atrophy does not extend above the lower third of the thigh, and in the upper extremities above the proximal portion of the forearm, atrophy and paralysis advancing in parallel. The hand frequently presents the claw type, while there are special deformities in the feet due to the development of paralytic club foot and the aspastic weakening. Changes in electrical responses are both quantitative and qualitative, and vary from slight reduction to complete absence, and occasionally show partial or complete reaction of degeneration. Disturbances of sensibility are common but not constant. Fibrillation is almost invariably noted. The patellar reflex is reduced or abolished, very exceptionally increased. The nerves in the affected parts show a progressive interstitial neuritis, but the lesion is not confined to the periphery as in the two sectioned cases which he admits to his classification the columns of Burdach were degenerated without the involvement of Lissauer's zone.

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The pyramidal tracts, the cells of the anterior horn, and Clarke's columns were involved in the degenerative process. In his own case the changes in the peripheral nerves were very slight, while the case of Marinesco, quoted by him, showed extensive neuritic changes. It should be added that in unusual cases facial, ocular, faucial and laryngeal muscles have been affected, and in certain cases the atrophy has progressed to the involvement of every skeletal muscle. Rarely optic atrophy has been recorded.

Since the appearance of Sainton's thesis a considerable number of additions have been made to the casuistics of the subject and the importance of this type of progressive muscular atrophy as constituting a link between the so-called pure myopathies and the spinal type of progressive muscular wasting has been recognized on every hand. However, some question as to the hereditary character of the disorder has been raised, and Ansbach even questioned whether it is hereditary in any way whatever. (*Univ. Med. Mag.* Vol. XIII. p. 287.) I have found in a revision of the cases published during the past eight years a notable family group reported by Warrington (*Lancet*, 1901, Vol. II, p. 1574). In this instance a family presented three cases of the peroneal type occurring in the mother and two sons. A more important instance is published by Brasch. (*Deutsch. Zeitschr. f. Nervenheilk.*, Bd 26, p. 302.) It concerned a family presenting the disease in three successive generations. In the first generation isolated paralytic club foot appeared, while in the third generation the club foot was attended by progressive paralysis and atrophy. In the second generation father and son presented weakness in the legs which progressed steadily to complete paralytic club foot. Commencing in the legs at the 14th year, at the age of 40, it was followed by involvement of the hands and arms. It then advanced with great rapidity, eventually presenting external characteristics similar to those of the Duchenne-Aran type. These cases presented widely distributed fibrillary twitching. The distal portions of the limbs were notably anesthetic in all four extremities. The reflexes were abolished, and there was qualitative change in the electrical responses of nerves and muscles. Pain and sphincter troubles were absent. Fabian (*Inaug. Dissert.*, 1898) also reports an instance in which a father and son were identically affected, as was also the father's brother.

I have recently encountered a case which presents, if we can rely upon the statement of the patient, a most notable instance of heredity. An attempt will be made to verify as many of the facts here recorded as is possible. It appears, however, that the disease has positively appeared in five successive generations, is perhaps apparent in the sixth, and there is unverifiable but reasonably dependable hearsay evidence that it was also present in three anterior generations in succession, making nine generations all told.

CASE.

PERONEAL TYPE OF PROGRESSIVE MUSCULAR ATROPHY OF THE FAMILIAL TYPE, EXTENDING THROUGH SIX AND PROBABLY NINE GENERATIONS

George T. W., 34 years of age, unmarried, was born in Cumberland, Md., and had three brothers and two sisters. Of these children, the oldest brother died in infancy. Two brothers survived. One, two years younger than the patient, is living somewhere in California, exactly where is unknown, and is similarly affected. The other, as far as known is well. His two sisters are living; one has been married six years but has no children, the other, who presents some weakness of the ankles, has a boy of ten who also has some weakness of the ankles and shows a tendency to stumble and fall in running and otherwise to call to the attention of the patient many of the features with which he himself is familiar in the case of his brother and in his own experience.

His mother's family consisted of four children, three sisters and one brother. The uncle developed the disease in early adult years and died at the age of 27, unmarried. Both aunts were married but had no children. Both are now blind, said to be affected with optic atrophy, and one is deaf.

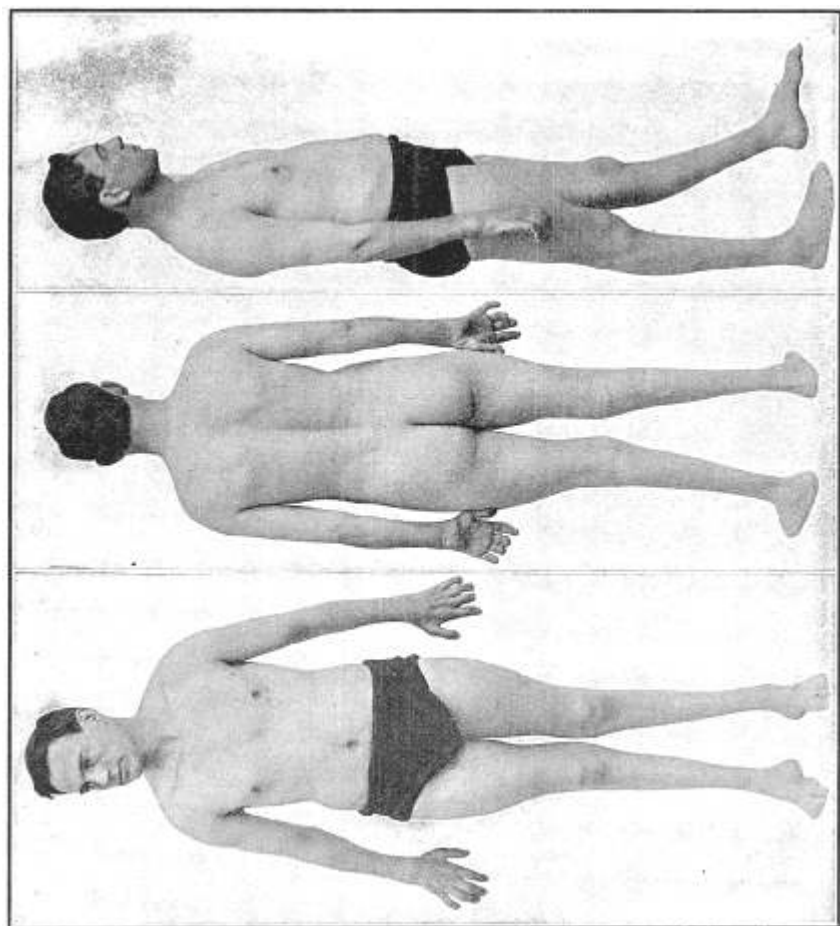
His grandmother on his mother's side was one of five children, having one sister and three brothers, two of whom were affected with the disease, and from these grand uncles most of the information is obtained regarding previous generations. One, who lived to reach the age of 70 was unmarried and even to the end of life was able to get about and do a little work, the affection being mainly confined to the lower extremities. The other was married and had 5 children, who with their children also are unaffected. The third grand uncle had seven healthy children.

The great-great-grandmother was also one of 5 children, having two sisters and two brothers, one brother being affected with the disease.

The grand uncles before mentioned insist that they were informed that three other anterior generations had shown instances of the same disease, in males only.

Owing to the fact that the disease has invariably descended by the female side with a change of family name in each generation, tracing is difficult. It is the belief in the family that the disease has tended to become more severe and more extensive with succeeding generations. Thus in the generation of the grandmother those affected were only partially incapacitated. In the patient's generation general disability is almost total; it appeared early and rapidly involved all four extremities.

Personal History.—The patient's parents separated in 1880 and three of the older children, including the patient, a brother and a sister, were placed in an orphan asylum in Auburn, N. Y., the mother taking care of the two younger children, the father going West. The patient has never had any serious illness, accident, injury, infection, or intoxication and his habits have been fairly good. In 1882 he was taken out of the asylum by a party with whom he stayed for a year, when he went to work as a newsboy and bootblack until 1885. His father, who had gone to St. Louis, sent for him to join him at that place, where he worked in the St. James Hotel as a bellboy until 1888. His ankles then began to bother him and running up and down stairs became difficult owing to the weakness of the ankles which were very apt to turn under him and throw him heavily. In 1889 the ankles were swollen badly. He was given a liniment for this swelling and a plaster cast was applied by a doctor in Belleville, Mo. A few years later he was advised to wear braces on account of the turning of his ankles which were then so badly affected that he was constantly tripping and falling over things. The swelling and pains disappeared, but he felt numbness in the feet and ankles, especially at night. Braces were worn for three years and finally left off on account of their weight. In 1891 he returned East and found some relatives living at Syracuse, N. Y., and then for the first time learned of the hereditary nature of his disease. From 1893 to 1895 he worked in a machine shop and while there noticed swellings in his hands similar to those that had appeared in the feet, but also observed that the hands would get numb and cramped when he attempted to do certain tasks, such as grinding a drill, and that at the same time his thumb and finger would become weak. He noticed that in picking up small articles his fingers were numb and that he would drop articles unintentionally. A physician in Syracuse sent him to a hospital, where he remained three months, receiving massage and electricity. He was discharged as incurable and advised to go to the country. This he did and worked about at odd jobs with sufficient ability to make his expenses. On advice he got a bicycle in order to exercise the muscles of the lower extremities. He used this for two years, when, owing to leg weakness he had to give it up. In 1898 he went to Syracuse and took up house painting; he had at different times worked as a



Showing Atrophy of Muscles in Distal Portions of the Four Extremities.

carriage painter but did not come in contact with lead in any manner until after the development of his disease. He was able to do such work as could be reached while standing, but was unable to mount ladders. This he continued until 1904, since which time, owing to advancing disability, he has been drifting about the country.

Status præsens. Upon examination we find a man of fair intelligence, about 5.11 in height, assisting himself with a crutch in walking, but with that aid getting along very briskly. Upon being stripped he presents a notable atrophy of the muscles in the distal portions of the four extremities. This is well shown in the photographs. In the lower extremities the small muscles of the feet seem to have practically disappeared, and he has no motor control of ankles or toes, the foot being flail like at the ankle joint. The muscles of the leg have all practically disappeared. There is a decided thinning in the muscles of the lower third of each thigh, particularly sharp in contrast with the notable adjoining muscular masses of the upper part of the thigh. The circulation in the parts is not very good and they are somewhat blue and cold. In the upper extremities the left hand is more involved than the right and shows a typical anthropoid position of the thumb. There is complete atrophy of the thenar and hypothenar eminences and excavation of the intermetacarpal spaces. There are no contractures anywhere, all joints being perfectly flexible passively, and a certain amount of power remains in the hands and wrists. The forearms are decidedly thin below the elbows. At the shoulder and pelvic girdle and on the trunk the musculature is excellent. There seems to be no local hypertrophy or pseudohypertrophy at any point. The facial, faucial and laryngeal muscles are intact and there is no evidence of disturbance of the cranial nerves.

In the atrophic parts reflexes as a rule are absent. Percussion on the extensors of the wrists and fingers gives reduced response. Knee jerks are present, heel jerks and plantar reflexes are entirely absent. There is no sphincter involvement and all bodily functions are well performed.

Sensation in the affected regions is notably blunted. The toes and feet are almost completely anesthetic, the legs show reduced sensibility for all forms of stimulation, the sensory disturbance ending at the knees. The same condition is present to a less degree in the upper extremities below the elbows.

Below the knees it is impossible with any current the patient can tolerate, and owing to the anesthesia a very strong one may be employed, to secure any response in muscle and nerve. In the muscles of the forearms and some of the muscles of the hand there is a partial reaction of degeneration. In some instances the response to faradism is very slight, while the muscles will respond to galvanism. Usually at such points positive and nega-

tive responses are equal, rarely the negative is the most active. Fibrillary tremor can occasionally be seen in the atrophic parts, and the patient now frequently experiences quivering of the muscles of the upper part of the thigh, the sternoclydomastoids and the eyelids. Owing to the weakness of the ankles where paralysis is complete the patient walks with a high knee action. By over-extending the knee and depending upon the ligamentous control of the ankle joint he is able to get along in a remarkably satisfactory way in view of the extent of the paralysis. In standing he sways about a good deal, balancing himself by the action of the hip and thigh muscles, but stands with eyes closed nearly if not quite as well as with them open.

According to the patient's best knowledge no female has ever been affected, but in this connection we may recall the interesting statements of optic atrophy in his two maternal aunts and the weakness of the ankles in his sister's case.